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Letter to the Editor

Fragile X Syndrome in Incestuous Families

To the Editor:

Reed [1954] suggested the investigation of children from incestuous unions as a method for calculation of the detrimental heterozygosity of man. Some studies of latent genetics load in man have been based on the comparison of health status of incestuous children with their half-sibs born to the same mothers in matings with nonconsanguineous partners [Adams and Neel, 1967; Seemanová, 1971]. These studies were limited to the detection of autosomal-recessive genes leading to abnormal phenotypes or mental deficiency in homozygotes. The highest coefficient of inbreeding in human beings is ¼ in offspring of incestuous matings: hence, the high proportion of affected homozygotes and low incidence of affected individuals among their maternal half-sibs. Mental deficiency in incestuous children represents not only cases of simple recessive inheritance. Recently, we observed three incestuous families in which fragile X syndrome was detected. The pedigrees are shown in Figures 1–3. The fra(X) children were born to carriers from incestuous unions as well as to un-

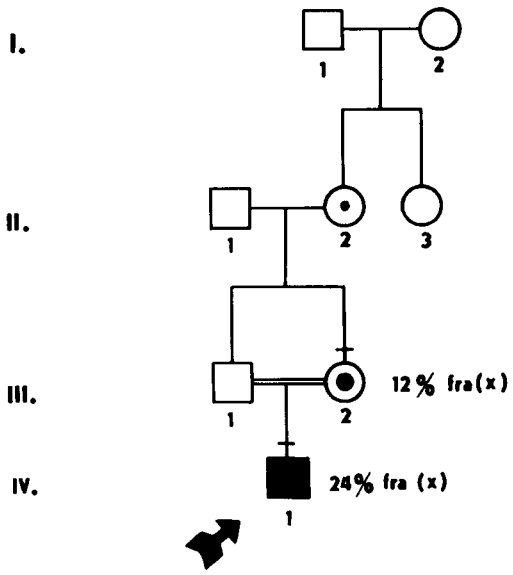

Fig. 2. Pedigree of family 2 shows brother-sister mating and its product, affected with fragile X syndrome.

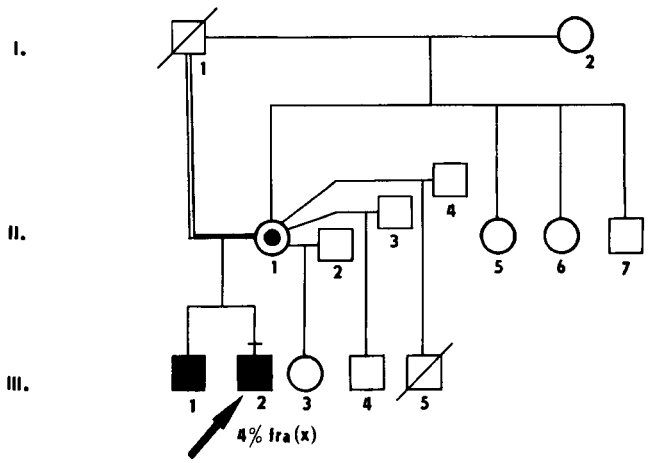

Fig. 1. Pedigree of family 1 shows two products of a father-daughter union, affected with fragile X syndrome, and three other unrelated partnerships of the incestuous mother, from which 3 unaffected children were born.

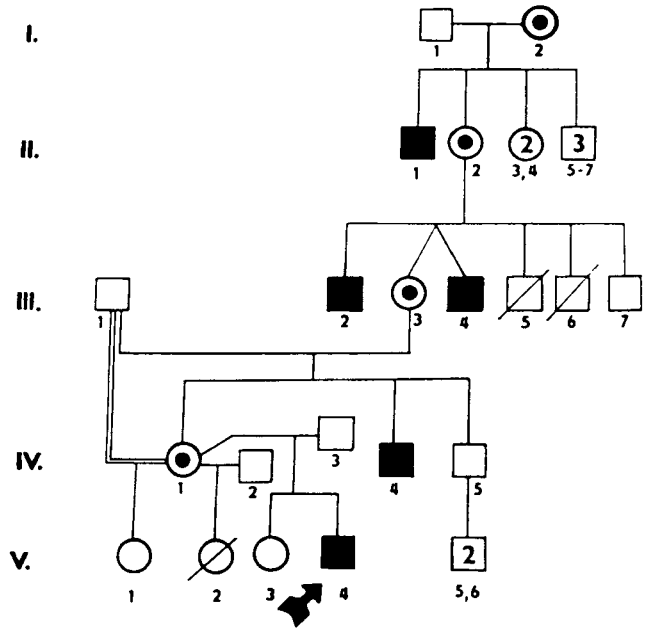

Fig. 3. By chance a healthy girl (free from fra(X) mutation) was born to the incestuous union. An affected boy was born from the unrelated union (control group of half-sibs of incestuous children).

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related partners. Therefore, we recommend use of incestuous children and their maternal half-sibs as a control group for studies estimating latent genetic load after investigation for fra(X). The incidence of fra(X) syndrome is high, and mental retardation in heterozygotes is uncommon. Both of these factors can play a role in the occurrence of incest, and in pregnancy at young age, as well as in multiple partnerships. Families of heterozygotes for fragile X should be excluded from the material for the calculation of human latent detrimental (autosomal-recessive) genetic load.

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